Phil Pallister MD

PUBLIC HEALTH, WELFARE & SAFETY

Exhibit No. 2

Date 1-31-07

Bill No. SB 162

For testimony on newborn screening

In 1973 I prepared a Chronological Brief Regarding Biochemical Screening of Newborns As It Pertains to Montana which outlined the whole business, and that **we were then screening for twenty-three genetic diseases** (fifteen amino acids, six mucopolysaccharidoses, & two sugar disorders) cheaper and more accurately than the state was doing for PKU alone and that we could screen for T3 & T4 (two thyroid tests) for an additional $3.43 per patient. Nine of the AA and sugar conditions were treatable then, some of the MPS disorders were possibly treatable while eight or nine of the thyroid conditions were all treatable. In addition RH and blood type conditions and rubella problems could be prevented. The state had missed about seven genetic disorders from 1959 to 1965 that we knew how to screen and to treat.

We had a list of all the cases identified:

PKU	-	34	
? Homocysteinuria	-	2 *	
? Hyperprolinemia	-	1 *	
Mucopolysaccharides	-	8 *	
Hypothyroidism	-	18 *	
Rubella syndrome	-	14 *	
Kernicterus	-	15 *	*All of these patients were in Boulder

It was costing the state $35,000 a year to treat and care for such patients then and their life expectancy was about 50 years. We could do all of these tests for $4 per patient on the 11,00 Montana newborns - $45,000 per year. We had done over 2000 such newborns in Boulder and Helena to demonstrate the efficacy of screening. I don't know the actuarial figures today but they are certainly proportional.

Last year I looked at the list of retarded (developmentally disabled – whatever that means?) patients in Boulder. There were 79. Only 21 carried a diagnosis. There was no admission screening, chromosome lab or genetic lab, etc. Some of the missed patients are scattered around the state in other settings.

There are at least two states screening for 50 disorders; Mississippi is one. Montana once led the nation. Now it is near the bottom!

Today the situation is much the same. We are missing patients and not identifying families that can be helped! It is costing us dearly!

Finding and treating one patient a year is sound economics for those interested in money.

I prefer to think of the social and human wastage that occurs from our neglect.

Newborn screening in Montana

1934 The first "inborn error of metabolism" – phenylketonuria (PKU) was described by Fölling, a Norwegian chemist – he published 11 cases.

1940 Jervis, Letchworth Village, Thiells New York, proved PKU was a recessive disorder inherited from both parents; in 1947 he demonstrated these patients could not metabolize an amino acid essential for brain formation (phenylalanine); in 1953 he demonstrated these patients had an inactive enzyme, phenylalanine hydroxylase, in their livers so they could not metatabolize and use phenylalanine. **He proved that the problem was inherited from each parent as a defect in metabolism and located the problem in a liver enzyme.**

1956 Three years later I met George Jervis in Richmond, VA, returned to Boulder where we screened 500 patients using Fölling's method, dropping a few drops of 10% ferric chloride in acidified urine, or on a diaper, leading to a deep blue color in PKU patients - the "diaper test." There were four patients in Boulder (two were sisters), four in their relatives, three on the waiting list and one in Warm Springs State Hospital for the insane – twelve in all.

Armstrong and Tyler at the new medical school in Salt Lake determined the brain damage was mostly caused by high levels of un-metabolized phenylalanine in the blood and were using a diet low in phenylalanine created at the U of Indiana called the Basal Mix diet. We began to treat a patient from Flaxville with their help. Her resulting IQ was in the sixties. Four of her relatives with PKU were less than IQ 30. The Ketonil diet came out in '56 followed later by Lofenalac

1959 In Hartford, CN, as a Fellow of the American Association on Mental Deficiency (AAMD, now called AAMR and soon to be AAIDD) and on the five member planning committee, **I proposed we push for national screening using the diaper test after the newborn has had one meal or more of milk**. The other physician on the committee, very senior and head of the Faribault, MN institution, blocked it – "since no treatment was available." **I returned to Montana and as the only GP on the Montana Medical Association's Maternal and Child Health Committee I made the proposal we screen in Montana: <u>the five pediatricians and the five obstetricians</u> voted against it using similar reasoning.**

Al Miller (Helena pathologist) and I began to do free of charge newborn screening at St. Peter's Hospital and Boulder and helped other hospitals set up their programs. Most of the testing was in Boulder

In the meantime 1/10-12,000 newborns in all of Montana were being missed!

Legislative Efforts

Very few physicians and others were interested in screening newborns for these rare diseases but **in 1965 the Butte Parent's group, with our stimulus, had SB 128 introduced by Senator R. T. O'Neill from Roundup with Reardon from Butte, Dessault from Missoula and others and it became law** (Chapter 108, 39th Legislative Assembly). The law was on the books but bureaucratic failure led to no oversight and little statewide testing occurred.

In Dillon, two successive infants with PKU were born to a young dentist and his wife in the Barrett Hospital and were finally diagnosed by Roger Clapp, a Butte pediatrician familiar with our efforts. Doctors around the country were being sued for not doing follow-up family studies, etc. and the dentist contemplated suing the state Department of Health but he moved to California.

1971 **Greg K. was born in Helena, tested positive, and was under treatment on his third day of life, several days before the state screening test was reported from Oregon. His mother told me his latest IQ was 117.**

1973 **Dr. Miller and I were testing for 24 disorders** and I had recognized that **hypothyroidism was as large a threat in Montana as PKU**, had set up several methods for testing for that disorder and was convinced we could test newborns. **Dick Welch, a pediatrician at the Department of Health and Chief of the Maternal and Child Health Division, and I sat in the dining room at the Boulder institution's hospital in the fall of 1972 and wrote three bills to enlarge our screening program state wide.** They were introduced by Gary Marbut from Missoula and others.

The newborn screening bill first included an **advisory committee** of a parent, pediatrician, geneticist and two others but was removed by the Legislative Council before introduction as not in accord with the new 1972 constitution. John Anderson, MD, head of the Department of Health, said he would form such a committee but he "dragged his feet" -his statement - for over nine months.

David Lackman, a Ph D microbiologist and head of the Department's lab, had vigorously opposed the idea of screening for hypothyroidism from early in 1972, had solicited many national leaders, from Massachusetts and NIH and others, for their supporting views, circularized each doctor in the state with these documents supporting his belief, and as late as April of 1974, Lackman was still avoiding implementing a test for hypothyroidism when he circularized (11 Jan 74) the opinion that their advisory group felt that a newborn test would be negative and should not be done unless there was jaundice for more than 4 days. Most cretins are not jaundiced that early, some not for months. At that time our law required the head of the lab be an MD pathologist; when I pointed this out the law was changed to accommodate Lackman.

Robert Guthrie of the Guthrie tests for PKU and other disorders, as a reaction to Lackman's continuing opposition to hypothyroid screening, called me re a letter of Lackman's asking for Guthrie's opinion. Guthrie cited two places (eg Montreal) where it was working and then he, geneticist John Opitz from Wisconsin and myself met with department personnel (Anderson, et al) as late as 9 Jul 75 and seemed to get nowhere. I do not know when the state finally picked up on the program to stop the development of cretinism in our newborns, but it was sometime later.

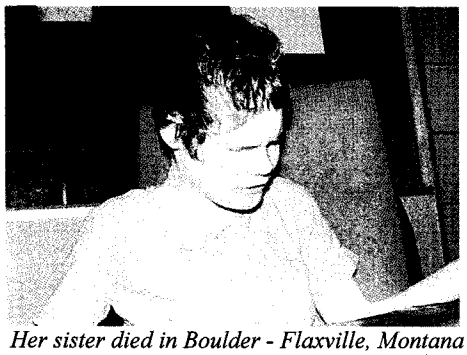
With this background it is obvious that I do not trust many bureaucrats, especially senior ones, and even some physicians to do what is right and honorable, to act as the scientists and doctors they should be. I moved from the state job in Boulder, started the Shodair genetics unit and was there for six years when I finally retired.

Last spring, much to my chagrin, dismay, watching the Today show, I learned that **Montana was screening for only four disorders although hypothyroidism was one**. What had happened to the 24 tests we had developed and left for them? Al Miller had demonstrated we could do it for about $1.60 per patient with another $3.60 for two hypothyroid tests? Jack Casey, now the Shodair Administrator, is also a registered medical technologist and was running our lab in Boulder and doing the testing. We were leading the nation!

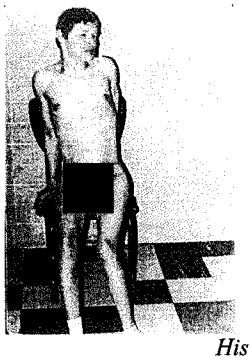
Now we have a new chance to take our place with the other informed and progressive states. Two are screening for fifty. Many persons still do not believe in testing if there is no good treatment (as was once the case for PKU), but this forsakes those families that need counseling after the birth of one affected child who are often not diagnosed for many months. We have missed kids with these disorders and for this I am sorry and I feel a need to apologize for my dereliction of responsibility in this area. The legislators do not need to apologize; they have always done their part in giving us the law. Those old bureaucrats should be ashamed of themselves.

Nevertheless, a new breeze is blowing and this bill is the result of modern health educators and leaders, nationally and in Montana, who have worked with an excellent group of advisors to bring us into the middle ranks, at least, in this area. I personally wish we were the nation's leader once again!

A few patients with disorders that are amenable to screening.



Her sister died in Boulder - Flaxville, Montana



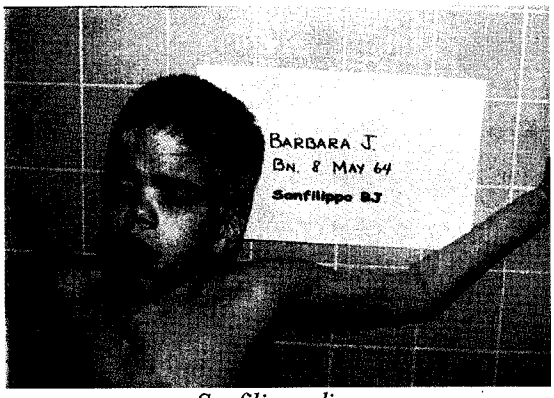
His affected sister born in Oregon

There were 14 Untreated PKU patients in Boulder in 1973

By 1973 we had diagnosed 34 patients with PKU from Montana families – at least five had been treated.

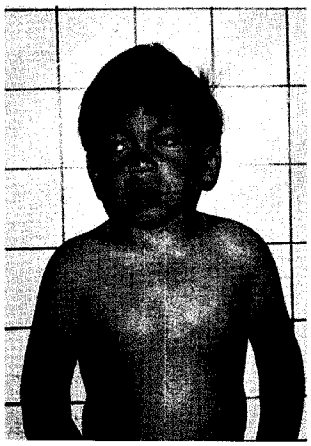
This is a treated Montana patient. His IQ is above average (117 by one measure)
Helena, Montana

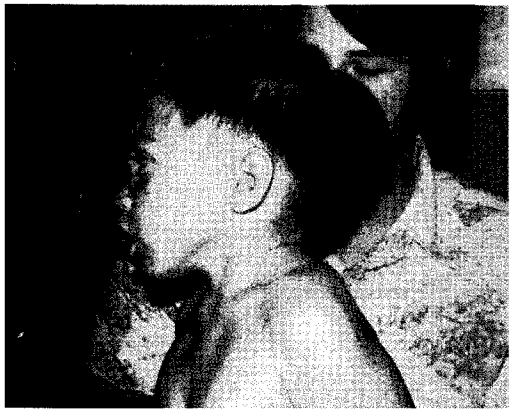
Sanfilippo diagnosed 10 yrs. postmortem
Rocker, Montana



Sanfilippo disease
Anaconda, Montana

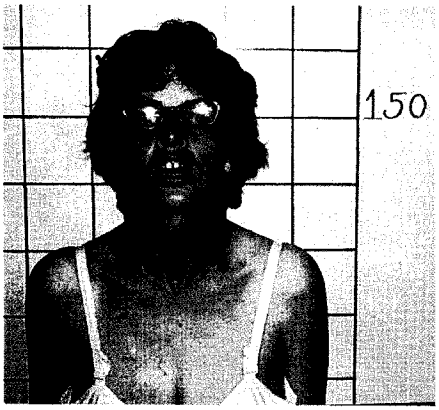
The AMPS diseases are not successfully treated at this stage of the game (just as PKU was back in the fifties) **but these families do need counseling as soon as the diagnosis can be made and some treatment is available.**



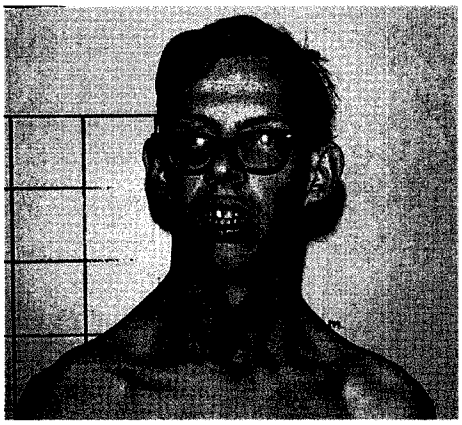


Hunter disease – an x-linked recessive storage disease - AMPS

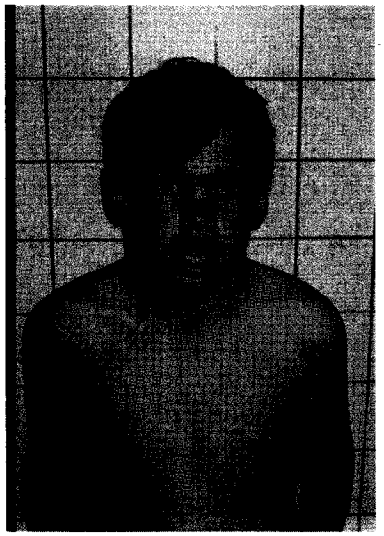
Hyperprolinemia-aminoaciduria



Sister of Jackie – cretinism, cancer of thyroid, frozen hands & feet

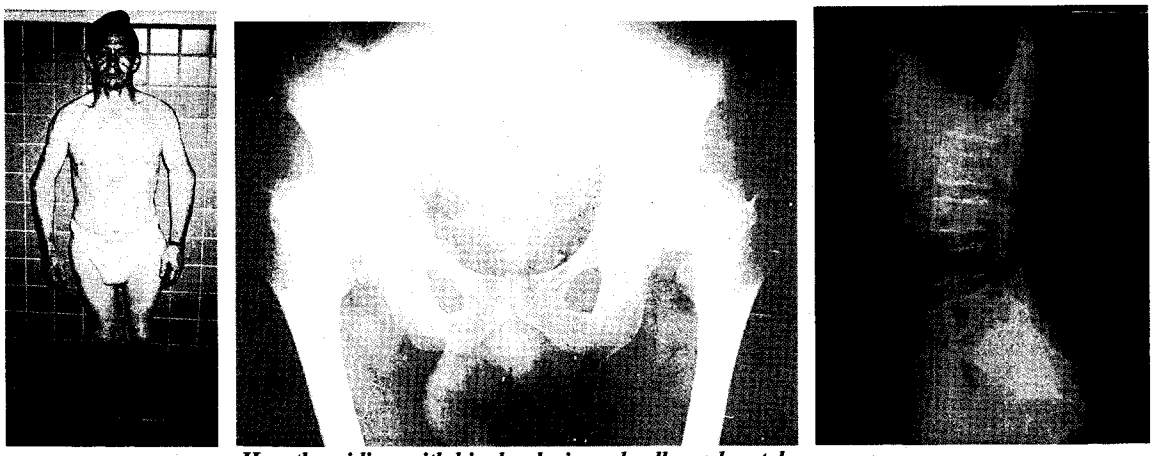


Robert- cancer of thyroid, cretinism

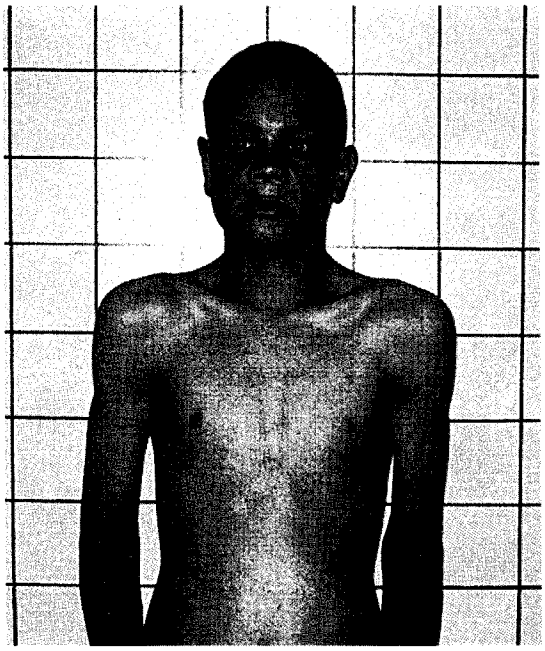


Jackie S. At age 11 he had an adenocarcinoma of the thyroid. These three siblings were affected. There were two normal siblings.

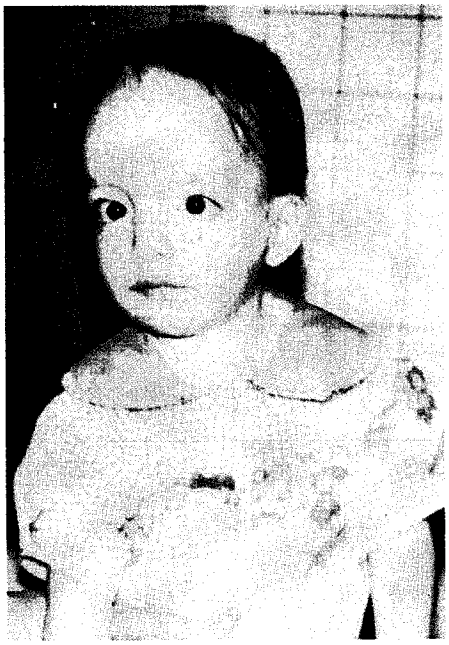
There were 18 untreated hypothyroid (Cretins) patients in Boulder in 1973



Hypothyroidism with hip dysplasia and collapsed vertebrae



Erythroblastosis fetalis with kernicterus (brain damage)
This is a blood type problem



Rubella (German measles)

In 1973 there were 14 patients in Boulder as a result of rubella. Last year there were none in Boulder. The vaccine has been effective as well as screening prospective mothers. There is a very reduced need for such screening now, although occasional outbreaks (200 cases north of us in Canada two years ago) demonstrate we should still exercise concern in this area.

There may be other tests besides those required in your state. More helpful information is available by contacting:

The National Newborn Screening and Genetics Resource Center
(512) 454-6419

www.genes-r-us.uthscsa.edu

or
call Montana's state health department's Newborn Screening Program
at (406) 444-1216

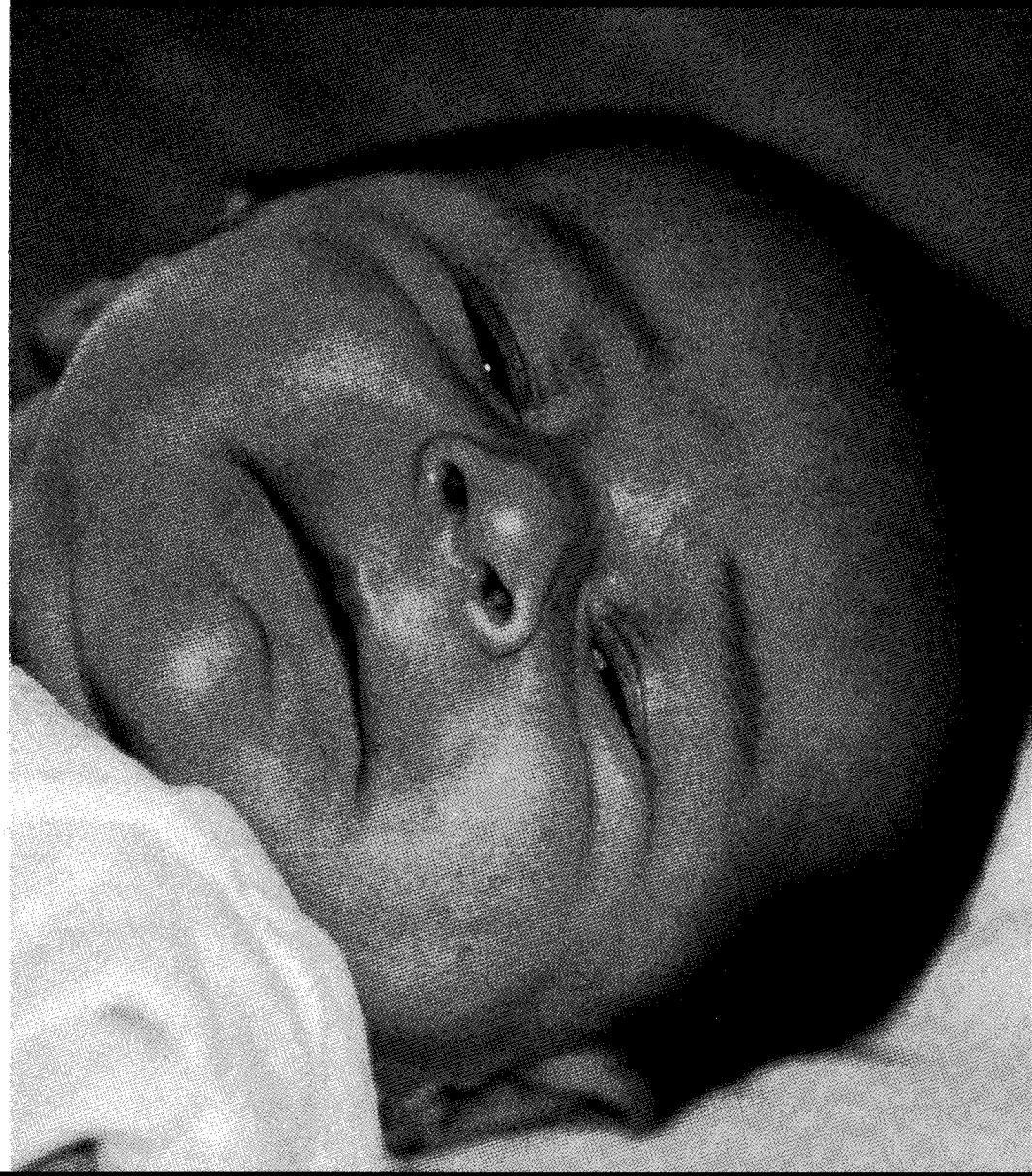
MONTANA
Department of Public Health & Human Services

600 copies of this public document were published at an estimated cost of $.10 per copy, for a total cost of $60.00, which includes $60.00 for printing and $0.00 for distribution.

This brochure was paid for from Montana's federal Maternal and Child Health Block Grant funds, grant # 6B04MC04233-01.

These Tests Could Save Your Baby's Life

Newborn Screening Tests



Why does my baby need Newborn Screening tests?

Most babies are healthy when they are born.

We test all babies because a few babies look healthy but have a rare health problem.

If we find problems early, we can help prevent serious problems like mental retardation or death.

How will my baby be tested?

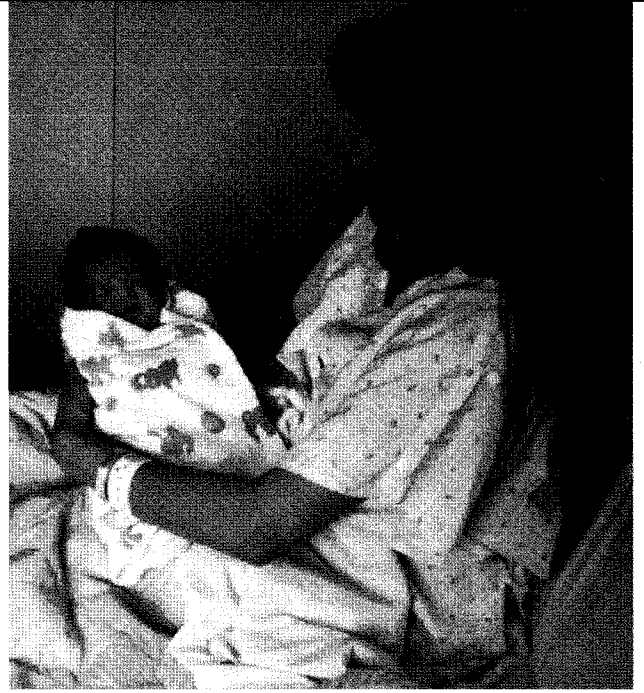
Before you leave the hospital, a few drops of blood will be taken from your baby's heel.

The hospital will send the blood sample to a newborn screening lab.

How will I get the results of the test?

Your baby's health professional will notify you of test results if there is a problem.

Ask about results when you see your baby's health professional.



Why do some babies need to be retested?

Your baby may be retested if you leave the hospital before 24 hours.

Some babies need to be retested because there is a problem with the sample of blood taken from the baby.

A few babies need to be retested because the first test showed a possible health problem.

What if my baby needs to be retested?

Your baby's health professional will contact you if your baby needs to be retested. They will tell you why the baby needs to be retested and what to do next.

If your baby needs to be retested, get it done right away.

Make sure that your hospital and health professional have your correct address and phone number.

What if I have questions?

Ask your baby's health professional if you have questions or concerns.

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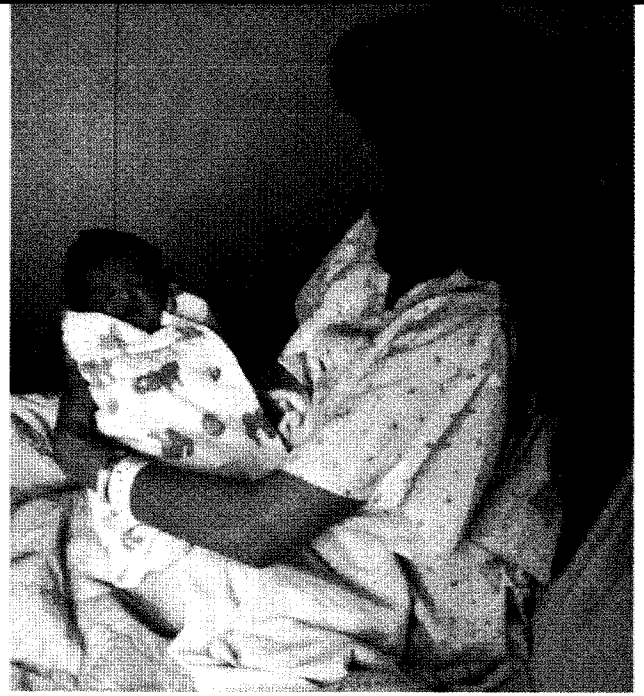

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January, 2007		Expected	
	Annual	In MT @	
	Expected	11,575*	Type of
NBS Condition	**Occurrence +**	**births/yr**	**Intervention**
Currently Mandated by DPHHS			
Phenylketonuria (PKU)	1/10,000-15,000	1	Dietary
Galactosemia (GALT)	1/40,000-60,000	0	Dietary
Congenital Hypothyroidism (CH)	1/3,000	4	Hormone replacement
Hemoglobinopathies - sickle cell anemia (Hb SS)	1/15,000	0	Prophylactic penicillin and immunizations
	1/400 in African		
	Americans		
Proposed screening to also be Mandated **			
Deaf or Hard of Hearing	1/1,000	12	Hearing aids, speech development, cochlear Implants
Cystic Fibrosis (CF)	1/2,500	5	Respiratory therapy, pancreatic enzyme supplementation
Congenital Adrenal Hyperplasia (CAH)	1/12,000	1	Cortisol or analogs
Biotinadase deficiency (BIOT)	1/60,000	0	Biotin supplement
12 Fatty Acid Oxidation Disorders	1/9,300	1	Dietary
14 Organic Acidemia Disorders	1/53,000	0	Dietary
5 Aminoacidopathies:			
Maple Syrup Urine Disease (MSUD)	1/53,000	1 in 5 yrs	Dietary control of protein intake
Homocystinuria (HCY)	1/100,000	0	Dietary and Vitamin supplements
Citrullinemia (CIT)	1/60,000	0	Dietary
Arginninosuccinic Acidemia (ASA)	1/60,000	0	Dietary
Tryrosinemia (Type I, II, III) (TYR I, TYR II, TYR III)	1/100,000	0	Dietary
+ From **Hawaii's Practitioner's Manual**, 8th edition, 2003			
* Projected based on last three years' birth cohorts			
Specific cost data on the disease and/or death of Montana babies without early intervention versus those who were screened, diagnosed and treated			
early in their lives is not available. However, the Centers for Disease Control and Prevention (CDC) estimate that the lifetime cost to society for a			
person with mental retardation in the United States is $1.1 million (adjusted to 2006 values). If PKU is not managed adequately, infants have a			
93% risk of mental retardation			